ASH1L gene

ASH1 like histone lysine methyltransferase

Normal Function

The *ASH1L* gene, also known as *KMT2H*, provides instructions for making an enzyme, called lysine-specific methyltransferase 2H, that is found in many organs and tissues of the body. Lysine-specific methyltransferase 2H functions as a histone methyltransferase. Histone methyltransferases are enzymes that modify proteins called histones. Histones are structural proteins that attach (bind) to DNA and give chromosomes their shape. By adding a molecule called a methyl group to histones (a process called methylation), histone methyltransferases control (regulate) the activity of certain genes. Lysine-specific methyltransferase 2H appears to turn on (activate) certain genes that are especially important for development of the brain.

Health Conditions Related to Genetic Changes

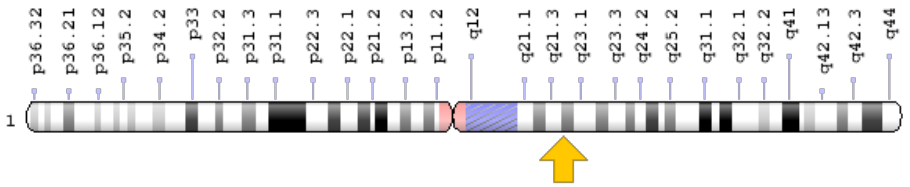
Autism spectrum disorder

At least seven *ASH1L* gene mutations have been identified in people with autism spectrum disorder (ASD), a varied condition characterized by impaired social skills, communication problems, and repetitive behaviors. Some *ASH1L* gene mutations associated with ASD change one building block (amino acid) in the lysine-specific methyltransferase 2H enzyme. Others delete genetic material in the *ASH1L* gene sequence or result in a premature stop signal that leads to an abnormally short enzyme. As a result of these mutations, the enzyme is nonfunctional. A lack of functional lysine-specific methyltransferase 2H enzyme disrupts histone methylation. The resulting changes in the expression of genes regulated by lysine-specific methyltransferase 2H affect brain development and increase the risk of ASD. Normal variations in other genes, as well as environmental risk factors, such as parental age, birth complications, and others that have not been identified, also affect an individual's risk of developing this complex condition.

Chromosomal Location

Cytogenetic Location: 1q22, which is the long (q) arm of chromosome 1 at position 22

Molecular Location: base pairs 155,335,263 to 155,563,160 on chromosome 1 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- absent small and homeotic disks protein 1 homolog
- ASH1
- ash1 (absent, small, or homeotic)-like
- ASH1-like protein
- ASH1L1
- histone-lysine N-methyltransferase ASH1L
- huASH1
- KMT2H
- lysine N-methyltransferase 2H
- probable histone-lysine N-methyltransferase ASH1L

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: Chromatin Mechanisms Regulating Gene Expression In Health And Disease
  https://www.ncbi.nlm.nih.gov/books/NBK45032/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ASH1L%5BTIAB%5D%29+OR+%28ASH1+like+histone+lysine+methyltransferase%5BTIAB%5D%29%29+OR+%28%28ASH1-like+protein%5BTIAB%5D%29+OR+%28ASH1%5BTIAB%5D%29+OR+%28absent+small+and+homeotic+disks+protein+1+homolog%5BTIAB%5D%29+OR+%28histone-lysine+N-methyltransferase+ASH1L%5BTIAB%5D%29+OR+%28huASH1%5BTIAB%5D%29+OR+%28lysine+N-methyltransferase+2H%5BTIAB%5D%29+OR+%28probable+histone-lysine+N-methyltransferase+ASH1L%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- ASH1-LIKE HISTONE LYSINE METHYLTRANSFERASE
  http://omim.org/entry/607999

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ASH1L.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ASH1L%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:19088
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:55870
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